ATM MUTATIONS IN BREAST CANCER

ABSTRACT OF THE DISCLOSURE

According to the present invention, there is provided a method of testing a subject to determine if the subject has a predisposition for developing primary or bilateral breast cancer which includes the steps of detecting a mutation in the open reading frame of the ATM gene (SEQ.ID.NO: 1) in a cDNA sample from the subject, in a genomic DNA sample from the subject, which mutation is selected from the group consisting of the mutations set forth in Table 4 and Table 5; or detecting a mutation in the mRNA corresponding to the open reading frame of the ATM gene (SEQ.ID.NO: 1) in a mRNA sample from the subject, which mutation is selected from the group consisting essentially of RNA complementary to the mutations set forth in Table 4 and Table 5, wherein the presence of such a mutation indicates that the subject has a predisposition for developing primary or bilateral breast cancer. Also provided is an isolated cDNA having a nucleotide sequence which differs from the sequence set forth in SEQ.ID.NO: 1 by including a mutation selected from the group consisting essentially of mutations in position 378 T->A, position 3383 A->G, position 1636 C->G, position 2614 C->T, position 6437 G->C, position 2932 T->C, position 2289 T->A, position 6096 A-> T, position 6176 C->T, position 6919 C->T, position 3925 G->A, position 6067 G->A, position 2119 T->C, position 1810 C->T, and position 4388 T->G. A marker for determining a predisposition for breast cancer is also provided.